



EDITORIAL

Michael Morgan

After the excitement of 1996 – the year of the FA gene – it might be thought that the pace of ataxia research in 1997 would slow down, that this year would prove an anti-climax. Far from it. Inside Georges Imbert reports on the latest scientific breakthrough into SCA 6.

If you've been following the scientific press you'll know that different research teams are at loggerheads over the function of X25 – 'The FA Gene' – even in fact if it is a separate gene. Inside we try to present both cases without favour.

Gene Therapy isn't something usually associated with FA, so it's exciting to hear of two attempts to introduce gene therapy from Australia.

Of course not all in *Euro-Ataxia* is about scientific matters alone. We are deeply conscious of the need to balance science – which may only benefit future generations – with articles on living with ataxia – to help ataxic people in the here and now. Inside we publish my personal perspective on the psycho-social aspects of the ataxic life. It's meant to start debate so all feedback is welcome.

Finally, Lunteren in the Netherlands is the location for this year's AGM – see inside for details.

IDENTIFICATION OF THE MUTATION CAUSING SPINOCEREBELLAR ATAXIA 6

Georges Imbert, PhD (IGBMC, Strasbourg, France)

Autosomal dominant cerebellar ataxias (ADCAs) are diseases that share a number of clinical and pathological features. Very recently (between 1994 and the end of 1996), the genes and mutations causing three different ADCAs have been identified. These disorders are the spinocerebellar ataxias of types 1, 2 and 3 (SCA1, 2 and 3).

It was not so surprising to discover that the mutations in all of these three ADCAs are in fact very similar. In each case, the gene that is mutated contains a repeat of the same genetic 'word' (a CAG trinucleotide) that is longer than the one found in the normal version of the same gene. For this reason, this type of DNA mutation has also been called trinucleotide expansion mutation.

The consequence of such an expansion is that the neurons of an individual carrying the mutation will produce a protein that contains in its structure a repeat of the amino acid glutamine, longer than the one present in the normal protein. It is indeed the presence in the neurons of such proteins with an excess of glutamine amino acids (called also polyglutamine expansion) that constitutes the first step of the process ultimately giving rise to the symptoms of ADCAs. Proteins with polyglutamine can be, for some as yet unknown reason, toxic for neurons.

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There are other neurological disorders like Huntington's disease and dentatorubropallidoluysian atrophy (DRPLA) that are caused by the same type of biochemical toxicity. As a consequence of the discovery of these mutations, it has been suggested that other types of ADCAs might also be caused by CAG trinucleotide expansions. Indeed, earlier this year, Zhuchenko and colleagues from the Baylor College of Medicine in Houston identified the mutation causing SCA6.

This team has analysed several hundreds of short DNA fragments containing CAG repeats and characterized their length in normal individuals and patients affected with cerebellar ataxias of unknown origin. They were able to prove that for one of these CAG repeats an expansion is present on chromosomes of a few patients, but absent on almost 1000 normal chromosomes. This finding allows therefore to define a new type of spinocerebellar ataxia: SCA6.

As in the other cases, SCA6 is caused by a CAG / polyglutamine expansion. There is however a significant difference between SCA6 and the other SCAs. In the latter cases, the role of the polyglutamine containing proteins in the normal functioning of the neuron is not known for the moment. On the other hand, the protein that is mutated in SCA6 patients is normally a part of a calcium channel, a structure found on the outside of neurons and that allows the entry of calcium in these cells. The polyglutamine expansion in this protein (called subunit alpha1A) could alter the normal functioning of the channel, resulting in the long term in the ataxic symptoms.

It is noteworthy that other types of mutations in the same SCA6 gene have recently been described. They result in either familial hemiplegic migraine or episodic ataxia, two other autosomal dominant neurological disorders.

In episodic ataxia (EA, also called hereditary paroxysmal cerebellar ataxia, HPCA), the mutations cause absence of expression of the calcium channel from the mutant chromosome, resulting presumably in reduced level (about 50%) of the protein.

In familial hemiplegic migraine (FHM), the mutations cause the production of a protein with a single amino-acid change, presumably leading to an abnormal functioning of the calcium channel.

In both episodic ataxia and familial hemiplegic migraine, features of spinocerebellar ataxia, such as cerebellar atrophy, degenerative ataxia and nystagmus, have been found in some patients, indicating some clinical overlap with SCA6.

A striking feature of the SCA6 expansion, when compared to other ADCAs, is its small size. SCA6 patients have 21 to 27 repeated glutamines, while SCA1 and SCA2 patients have more than 35 repeated glutamines and SCA3 patients have at least 60 glutamines. The high expression of the calcium channel in the

cerebellum and the ataxic symptoms resulting from non polyglutamine mutations may explain the higher sensitivity of the calcium channel alpha1A subunit to polyglutamine expansion.

The next step for researchers working on SCA6, and also on the other polyglutamine disorders will be to understand the succession of events linking the mutation (presence of an abnormal protein in the brain) and the disease. Due to the similarities between all these diseases, it is possible that they all share the same pathological mechanism. Therefore, if one of the growing number of teams working on polyglutamine disorders unravels this mechanism for one of the diseases, it could help understand quite a lot about all the others.

BACK TO LUNTEREN

Michael Morgan

The 1997 EURO-ATAXIA AGM will be held in Lunteren, The Netherlands, between 24-26 October. Perceptive readers will recall that this was also the location for the 1995 AGM. Actually the Flemish were supposed to host the event but problems intervened to scupper their plans. The Dutch then came to the rescue with another invitation to Lunteren. For those of us who were there last time it's a comfortable, spacious centre, set deep in the woods of central Holland. I couldn't tell you where it is either, nor how to get there, only if you're coming by plane, the thing to do is fly into Schiphol where you'll be picked up and transported all the rest of the way by coach.

The design of the AGM will follow the usual EURO-ATAXIA format. Friday 24 October evening sees the arrival of delegates, also the official opening of the 1997 AGM. Main business of AGM will be on Saturday 25 October. The morning session will be devoted to medical and scientific issues in ataxia research, and will be addressed by Michel Koenig, Ewout Brunt, a cardiologist and, if there is news on the research into FA hearing problems, Jan Feenstra.

As is customary the afternoon session will focus on living with ataxia. 'Communication' is this year's theme, and particular attention will be given to the mighty Internet and related means of electronic communication. The hype tells us it's now possible to achieve well-nigh instantaneous communication between continents. Well, now's our chance to go beyond the hype. We hope to have a computer and phone-line on hand to give a practical demonstration of what it means to be wired. Such wonders as the Internet and World Wide Web, e-mail and discussion groups such as INTERNAF – the 'online' international ataxia community – will be fully explained.

Furthermore Carolien Koopmans will tell us about her

life 'without and with computer', stressing the enormous possibilities a PC gave her, as well in her professional life (getting her doctor's degree) as in her private life (as secretary of the Dutch FA group).

Finally Ineke Roelofs, one of the driving forces behind the EAGS (European Association of Genetic Supportgroups) will address the meeting on the topic of European cooperation.

The final session, on Sunday morning 26 October will deal with the business side of this year's AGM. It promises as well to be a very busy session as a lot of key decisions have to be made.

First a new president must be elected to succeed Manfred Van den Kerchove who resigned suddenly and unexpectedly earlier this year. Also a new EURO-ATAXIA Board has to be selected. On top of that we hope to carry out an in-depth evaluation of EURO-ATAXIA – where it's going and why isn't it going where it should be – on this our eighth anniversary. On the financial side too, this year's AGM will be on a less secure footing. All delegates and accompanying helpers will have to pay their own travel expenses. All non-delegates and their helpers will have to pay accommodation expenses on top of that. It may be enough to make some wish to give it a miss and stay at home this year – but please don't. This year's AGM looks set to be a crucial one for the future of EURO-ATAXIA. For those of us who care about EURO-ATAXIA it's not to be missed!

AUSTRALIAN FA RESEARCH

Dr. Ian Alexander

It used to be thought that because Friedreich's Ataxia was in large part a brain disorder it did not present itself as a suitable candidate for gene-therapy research. However there are two such projects currently being carried out in Australia. Below Dr. Ian Alexander of the Gene therapy Unit in the New Children's Hospital, Sydney, Australia gives an overview of this research.

Now that the gene responsible for Friedreich's Ataxia has been identified and isolated, doctors and scientists can begin to study the gene and its function in greater detail. There will be two definite and immediate benefits: first a much greater understanding of FA, and second an improved ability to diagnose the condition using genetic tests. It can reasonably be expected that these benefits will be realised within a relatively short space of time, perhaps 2-3 years. A third potential benefit from the cloning of the FA gene, and the one most hoped for, is the possible development of improved treatments. Gene Therapy is currently the most promising possibility, although other approaches

may become apparent as the nature of FA becomes better understood. For those unfamiliar with the concept of Gene Therapy this would involve delivering normal copies of the FA gene to cells within the body affected by the condition. Although simple in concept this is a very difficult task to achieve. Cloning of the FA gene is certainly the first step along the road towards Gene Therapy for FA but there are many more formidable obstacles to be overcome. Foremost among these is the need to develop the technologies required to deliver the FA gene to the appropriate cells within the body.

Because FA primarily affects the nervous system we need to identify and develop methods for efficiently delivering genes to nerve cells. Our current approach is to use genetically engineered viruses for this purpose. Already we have identified a viral delivery system with special promise and we are now in the process of studying how useful this system will be in the possible treatment of FA by Gene Therapy. Although this represents exciting progress there is still a long way to go and it should be appreciated that it could be quite a few years before this type of research has a practical impact on the lives of people and families living with FA. In fact two major research programs into gene therapy for Friedreich's ataxia have been started during the past year in Australia. One of these is at the Murdoch Institute, in the Royal Children's Hospital in Melbourne, and the other at the New Children's Hospital in Sydney. The Murdoch team is led by Bob Williamson, the new Director, who helped to locate and identify the FA gene when he headed the FA group in London (with Sue Chamberlain). The Sydney group is directed by Ian Alexander, who has recently returned from a very productive three year stint with one of the best of the American gene therapy groups in Seattle. Both groups are trying to develop ways to put a copy of the normal gene into the tissues which are affected in FA and other conditions affecting the nervous system. Because of each group's previous experience, they are taking different but complementary paths – the Sydney team will try to use a virus as a 'Trojan horse' to carry the normal gene into cells, while the Melbourne group will start with the gene packaged in tiny fat particles.

In FA one of the major sites affected by the disease are the dorsal root ganglia (clusters of nerve cells) lying along the spinal cord. Effective gene therapy for FA will require the ability to efficiently deliver a normal copy of the FA gene to these sites in such a way that the gene exhibits normal and long-term activity. The 'Trojan horse' strategy employs the natural ability of a harmless virus to carry genes into cells. There are a number of these gene delivery systems under development and each has its strengths and weakness. The first bit of good news is that the FA gene will fit into the delivery system we are working with. We were

only able to establish this after the gene was found. At this stage we have not placed the FA gene into our system as we first need to explore how well the system will work in delivering genes into nerve cells. To study this we have constructed gene packages that tell cells how to make certain enzymes which can easily be detected in cells using simple staining procedures that give rise to colour (e.g. blue). In this way we can use our gene delivery system to deliver these gene packages into nerve cells and determine whether we have been successful by checking to see if our staining procedure will now turn the cells blue. Put simply, if a cell goes blue we know the gene got in and is working. The second piece of promising news is that we have tested our system on rat nerve cells growing in tissue culture and have demonstrated gene transfer. We are now performing additional studies using this experimental approach to improve the efficiency of gene delivery. We are also on the verge of delivering genes into dorsal root ganglia of live rats. These latter experiments will take at least another 6-12 months to complete. We were recently excited to learn that a group in the US has successfully delivered genes into the rat spinal cord using exactly the same approach we are developing.

This growing FA research effort in Australia is an exciting development that should be a source of hope and encouragement to all those whose lives are touched by FA. However, while we are both very pleased that 'the show is on the road' and we are making a start, we also have to emphasise that it is a long road. Because the problem is very difficult, the path is uncharted, and safety is paramount, we are looking at an effort which may take several years before we can even begin to think of clinical impact.

[Printed with permission of Dr. Ian Alexander, Gene therapy Unit, New Children's Hospital, Sydney, Australia]

HEREDITARY ATAXIA SCIENTISTS TO MEET IN CANADA

Montreal provides the setting for an intensive seminar on Hereditary Ataxia. The meeting will take place over the weekend of May 29 – June 1. There will be workshops on:

1. Clinical studies (including imaging and neurophysiology)
2. Genetics: mapping & cloning, population genetics
3. Triplet repeats (CAG & GAA), mechanisms at the DNA level
4. Polyglutamine proteins
5. Frataxin
6. Phenotype-genotype correlations
7. Treatment

A general introduction will be given by Dr. Fred Andermann the evening of the 29th, and a summary session will be held on the morning of June 1. The meeting will be at the Delta Hotel in Montreal. A session for lay people/associations is scheduled for the afternoon of June 1. It's estimated that there will be about 70 invited participants, and about 50 Canadian neurology residents will be invited to attend. It's a good way of presenting the subject of ataxias to neurologists in training.

Funding comes from Hoechst-Marion-Roussel Canada, from NAF, and is pending with MDA and the Foundation for Rare Disorders (NIH).

NEWS IN BRIEF

1997 will be a very important year for EURO-ATAXIA. The time has now come to make an in-depth evaluation of the organisation's performance at all levels. We need to get more involved in the developing disability politics in Europe, so EURO-ATAXIA has applied for full membership in the new EDF, European Disability Forum.

The Spanish National Ataxia Group (AEAH) has been dissolved. The group of Madrid (an autonomic group) thought to take over the membership in EURO-ATAXIA, but financially they can not afford the membership. They ask to be 'silent' members.

The Ataxia Group organised inside the Swedish Neurologiskt Handikappades Riksförbund has applied for full membership of EURO-ATAXIA.

We received a same request from the International Joseph Diseases Foundation, Inc.

Daniela Iser from Switzerland asked to be admitted as a individual member.

Meanwhile the editorial board of *Euro-Ataxia* has also been enlarged to four members: Michael Morgan, Carolien Koopmans, Hans Doré, Marco Meinders. Our new policy includes an aim to exchange the news which is published in the national newsletters. We can organise translations into English, but don't keep news for yourself. We ask you to send interesting articles of the national newsletters to Hans or Carolien, who are co-editors of *Euro-Ataxia*. The national delegates are charged with the distribution of the *Euro-Ataxia* newsletter to other persons than those who are on the EURO-ATAXIA mailinglist.

X25 or STM7?

Last year, 1996, was one of the most significant in the history of ataxia research. In March came the electrifying news that the Friedreich's ataxia gene had at last been located and the miscreant protein identified – frataxin. (Campuzano, V. et al., *Science* 271, 1423-1427, 1996). Then in the October 1996 edition of *Nature Genetics* Carvajal et al. stated that the frataxin gene mutated in Friedreich's ataxia (FRDA) and the neighboring STM7 gene form a single transcriptional unit, a phosphatidylinositol-4-phosphate-5-kinase (Carvajal, J.J. et al., *Nature Genetics* 14, 157-162, 1996). However the original researchers remain adamant that frataxin, not phosphatidylinositol-4-phosphate-5-kinase (PI4P5K) is defective in Friedreich's ataxia (Cossée, M. et al. *Nature Genetics* 15, 337-338, 1997). On which Chamberlain et al. replied that 'Cossée et al. have failed to present either a plausible explanation for our original observations or a definitive argument to contradict our interpretation of the data' (Chamberlain, S. et al., *Nature Genetics* 15, 338, 1997).

Because it deals with very sharp scientific arguments, EURO-ATAXIA as a lay organisation can not and will not take side. We expect more results to come out soon to settle the issue.

ATAXIA: THE PSYCHOSOCIAL DIMENSION

Michael Morgan

Degenerative ataxia is caused, not by one disease but many. There are at least twelve separate disease syndromes grouped together under the collective title of 'hereditary ataxia', and although many symptoms will be held in common, there is also wide variation between types. This variation extends into psychological response, in particular the development of self and personal identity.

The crucial period for the development of self is that between childhood and adulthood – adolescence. Ataxia which strikes the pre-pubescent child will elicit a markedly different psychological response than one which effects the adult self. Friedreich's ataxia is the best known early-onset type of ataxia, extending from childhood to adulthood. Life with FA-type resembles a career, where the ataxia is an in-built characteristic of self. Many cerebellar ataxias are, however, late-onset type and so the ataxia may be likened to other 'external' diseases such as cancer.

As well as variation between different types of ataxia, there is also variation within specific ataxia syndromes. It matters a great deal, in psychosocial terms, if a young person has been using a wheelchair since childhood, or whether they remain on their feet until

late adolescence, or early adulthood.

What follows then is an attempt to deal with the psychosocial aspects of the 'typical' developmental sequence of ataxia, from childhood onwards, even though in practise few cases of ataxia are exactly alike.

The first signs: diagnosis and after

The first intimation that 'something is wrong' may come long before any medical authority is even approached. Anxious parents will notice signs of increasing 'clumsiness' in their child (sometimes even for years) before a definite decision is taken to seek medical attention, most usually the family GP. The 'clumsy child' is then referred by the GP to a hospital consultant, normally a Neurologist, but sometimes an Orthopaedist. Diagnosis is confirmed only after lengthy examination (although ongoing developments in genetics promise to both speed this up and make diagnosis virtually 100 per cent accurate). It is the parents who are most affected by the news rather than the child itself, who may even enjoy being the centre of attraction as a medical celebrity. For the parents, however, the news can be devastating. Quite apart from the question of provision, trivial by comparison, there may be tremendous feelings of guilt and shame. Parents invariably blame themselves for what has happened, and although this is 'true' in a basic, biological sense, it misses the point that such an outcome was both unforeseen and unintended. Although genetics promises to deliver quick diagnosis and even quicker pre-natal testing in subsequent pregnancies, the family may already be complete before the first child presents symptoms. Termination of affected foetuses is thus rarely a practical option – never mind the moral and ethical dimensions. The possibility that other children in the family might also develop ataxia can in itself create untold anxiety for parents. Other siblings are closely watched, scrutinised and, above all, worried over. Even the 'normal' accidents or 'clumsiness' of childhood are picked up and re-interpreted in the light of a positive diagnosis of ataxia in one child. For parents it can be an exhausting, nerve-wracking business. Constant anxiety and stress therefore, is an ever present feature of life for parents.

The child with ataxia will be oblivious to all this – or seemingly so. In fact the child will pick up quite a lot through intuition – a sharply developed sense in childhood. First, the behaviour of parents towards the child will change, even if imperceptibly. A perceptive child will notice, though. Second, visits to hospitals become more frequent; celebrity status loses its appeal. As well, there may be a shift in response from other siblings within the family, or from others outside, a subtle variation in the channels of interpersonal communication that the child with ataxia in-

habits. Again, a perceptive child will quickly notice increasing difference from others, leading to a special, often privileged, status within and outside the home. This in fact is the origin of the 'disabled other' he or she is to become.

As well as the emotional shock engendered by a diagnosis of ataxia parents are apt to feel bewildered as to what to actually do. Much depends on the rate of progression. If fast, the child will become a wheelchair user within a relatively short time, will enter special education from an early date and will have thus already acquired a 'disabled' status. If slow, however, the child will remain outwardly healthy for quite some time, with only marginal deterioration; he/she will enter a normal educational environment, and so retain a 'normal' status for that much longer. This crucially effects the strategies which parents are liable to adopt. They may try to hide the true facts from their affected offspring, to maintain a pretence of normality; a strategy bringing with it as many risks as benefits, not least the continual strain of deception within the family this entails.

At the other extreme, a too early exposure to the realities of ataxia may be needlessly brutal, even if well-meant. Bringing the only slightly disabled youngster, at the start of his ataxic career so to speak, face to face with those in the later stages of the disease can be a traumatic experience – best avoided if possible.

National Ataxia Groups are a major source of help and social support, more usually to the parents at this early stage than to the youngsters themselves. The ataxias are, by any comparison, rare. Most parents will never have heard of them before and so may feel isolated and anxious, unsure what any of this means. For the youngster with ataxia, however, life may not as yet pose that many obstacles. Whilst other children can often be breathtakingly cruel (especially from an adult point of view) children envisage their world as a unitary one, wherein little formal distinction is made between disabled and able-bodied companions. Rather it's the next stage, adolescence, where the distinction between disabled and able-bodied becomes a major personal and social divide; where, properly speaking, the problems, much more emotional than physical, really start.

The teen years: ataxia and the adolescent

Puberty consists of physiological and hormonal changes within the bodies of young males and females, transforming them into fully functional sexual beings. This in turn causes the tremendous psychosocial upheaval known as adolescence: the transition from childhood to adult status. Adolescence is the 'dangerous bend' of existence, when the securities of childhood are left behind as the search for adult identity begins. Parental authority is rejected. Self is now defined through interaction with peers, critically

through sexual interaction. Teenage peer groups become important as an alternative source of authority. Outwardly anarchic, these informal groupings are in fact highly conformist. Rigid conformity (albeit to 'alternative' group norms) is maintained through the development of status hierarchies. Status reflects the self: for this reason it becomes an all-important resource for effective sexual interaction.

The adolescent's social world is thus fragmented into divisions of status, and one of the most basic is that between normal and disabled. Disability means low status almost by definition, exclusion from or marginalisation within one's peer group and, most crucially of all, limited opportunities (sometimes a priori rejection) for sexual interaction.

For the teenager with ataxia the transition from childhood to adulthood can be paralleled by a shift from 'normal' to 'disabled' status. At this, still relatively early stage, the youngster is caught between definitions. He/she exists within the 'normal' world of interaction with peers, sharing in the 'normal' activities, values and attitudes – including the negative ascription of 'the disabled'.

As the ataxia progresses however, the youngster will find it increasingly harder to maintain a 'normal' status, being pulled inexorably towards the 'disabled' category. On a purely physical level the teenager with ataxia may find it harder to engage in normal adolescent activities: going to rock concerts, friend's houses, etc. This in turn will lead to increasing exclusion from core activities within the group, particularly those involving sexual interaction, thus marking a general downturn in social and personal status. Desperate to avoid the collapse in social status that a self-definition as 'disabled' will bring, the ataxic teenager will experience his increasing disability with intense shame, a purely personal failure. Denial and camouflage become common responses. These strategies, however, get more difficult as the ataxia develops, but are rarely dispensed with altogether.

Loneliness is a great problem for the ataxic adolescent. There are a number of specific reasons (as well, of course, as 'normal' ones).

First, there is sex – or rather the lack of it. Many young people with ataxia feel lonely, not because of lack of same-sex friends, but because they cannot share in boy/girl relationships. Sexual interaction remains problematic. Of course pretty much the same problems in sexuality are faced by any young person, whatever their physical status, but having ataxia as well will, however, magnify and give a sharper edge to the 'normal' problems it must be admitted.

Second, many ataxic teenagers find it difficult to establish close friendships, as with a 'significant other'. Friendship develops by way of self-revelation, as a process of increasingly frank exchanges. This presupposes a shared background of knowledge and experi-

ence which makes such interactions meaningful. The point about ataxia is that it robs the interaction of equivalence and, so, depth. Ataxia is 'off the agenda' within such situations precisely because it is not a shared experience. An interactional vocabulary does not exist for it within 'normal' settings. Third, identity with other ataxic people, via a National Ataxia Group, is often rejected. The individual's sense of self remains rooted on his/her 'normal' experience and status. To admit, even to oneself, that involvement with a group of other ataxic people might be beneficial is to risk a self-definition as 'disabled', and this is something which cannot be accepted at this vulnerable stage.

There is however another point concerning identity and disability which perhaps should be mentioned here. Our analysis so far has focused on how the ataxic self attempts to preserve its 'normal' identity against encroaching disability. But there is also an often contradictory impulse among ataxic people, to attach themselves to a 'disabled' identity because it offers a definite identity and so removes the uncertainty of living between normal/disabled status.

The Adult Self: Ataxia And Acceptance

Early adulthood for the young person with ataxia brings major crisis, the most intense crisis of his/her life to date: the decision to use a wheelchair. 'Decision' is perhaps the wrong word to use here, as it implies choice. In reality there is none, or little.

Physically, the situation will have deteriorated to such a level where walking becomes difficult, even dangerous. Using a wheelchair substantially increases personal independence and mobility. Ataxic people in wheelchairs can cover the same ground quicker, and most importantly, independently, than somebody who continues to rely on others for support.

This, however, may mean little to the person already within the crisis. For him/her the central issue is not mobility but rather personal identity and self – its roots are psychological, not physical. Using a wheelchair means accepting a self-definition as 'disabled' – the very thing that the young person with ataxia has spent his/her adolescence desperately trying to avoid. Hitherto, emphatic denial of disability has been commonplace: 'I will never go into a wheelchair'. Now however, the crisis-point has been reached, bringing a changed reality in its wake. The new order, psychosocial as much as physical, must be adjusted to on its own terms.

Acceptance of disability is always problematic. At a simple, everyday level a person can readily accept being 'disabled' – 'I use a wheelchair, therefore I am disabled' – whilst rejecting the often negative, patronising treatment meted out to 'disabled people' in wider society.

However, there is also a deeper issue. While acceptance may be easy enough at an intellectual level –

one can even pride oneself on one's own sagacity and maturity in doing so – this may not be the case at the more basic emotional level of self. Within the human psyche intellect and emotions run on different tracks – 'the heart has its reasons which reason knows nothing of', to quote Pascal – and too much discordance between the two leads to psychological conflict. This inner tension frequently erupts outward into irrational behaviour, inward into deep depression and quite often, to both. Depression with ataxia invariably takes on a serious form, much deeper than daily 'blues', and more of a general collapse in motivation. Like Chekhov's Uncle Vania, the trivial rituals of everyday life seem intolerably heavy precisely because we have lost overall direction: we simply don't see the point.

The idea of suicide may also seem attractive at times. It offers the illusion of control, that the self remains in charge, despite what is happening to the body. In fact it's never a solution. Life with ataxia can certainly be difficult at times, but still worthwhile, and still capable of being lived to its limits. Life with ataxia is, above all, a process of continual negotiation, renegotiation and adaptation. It is also inherently stressful, and this may give rise, periodically, to dreams of suicide as a way out. But it's nothing of the sort. The idea of suicide may have gotten Nietzsche through many difficult nights, but even he had to rouse himself in the morning, get out of bed, let the dog out, make breakfast and do all of the other mundane things that make up everyday life.

In a sense the acceptance phase of ataxia may be likened to the bereavement process, whereby people come to terms with their loss through a succession of internal psychological stages: denial, anger, withdrawal, depression and despair. This acceptance phase of ataxia is unquestionably a heavy burden, even, at times, an overwhelming one. But we must also remember that the acceptance phase is a temporary one. It is a transitional state, and has an end as well as a beginning. Finally, ultimately, the self must re-emerge into a state of relative equilibrium, having learnt to negotiate with external reality rather than being crushed by it. It will be a stronger self too. For the interim though Paul McCartney's advice is probably best: Let it be ...

Adult ataxic people will also have to contend with much of the negative treatment meted out to disabled people in today's society: stigmatisation, lack of opportunity and widespread discrimination in employment are, unfortunately, endemic. Disabled people tend to be excluded from the 'micropersonal' world of daily contact wherein sexual contact, for example, is first made. The direct consequence of this exclusion may be to reinforce isolation and deliver blows to ataxic people's inner sense of self and self-esteem, themselves critical resources for further interpersonal interaction. Life with ataxia will never be easy, but neither should

it be impossible. The aim of Ataxia Groups at whatever level of operation – Local, National or European – is to help ataxic people everywhere fulfil their potential in life.

The mature years: late-onset ataxia

The ataxias, we have noted, divide psychosocially in terms of age of onset. Major differences in psychological response are discernible according to which onset-category the individual with ataxia belongs. ‘Late-onset’ usually means anything from age 25+. British broadcaster and journalist, Glyn Worsnip, was in his mid-forties when he first developed symptoms of ataxia, so even within the ‘late-onset’ category a wide variation is common. Most of the late-onset types will be cerebellar ataxia (although a late-onset form of Friedreich’s ataxia was also, until recently, classified by clinicians).

The key division between early-onset/late-onset types seems to be the experience of ataxia in the formative years of self, particularly during adolescence. Early-onset types develop an external coating of self, a resilience born of their internalisation of ataxic reality as ‘normal’ reality. In contrast, late-onset types experience life with ataxia in terms of loss; as subtraction from what has been. This, in turn, creates an added vulnerability and instability among late-onset types. This can lead to the seemingly paradoxical position whereby late-onset CA sufferers exhibit more emotional and psychological damage than often much more physically impaired early-onset FA sufferers.

Relationships are as problematic for people with late-onset ataxia as for early-onset types, but usually from a different perspective. Whereas the early-onset type may have experienced most problems in making and sustaining sexual relationships in the first place, the late-onset type may well experience problems in existing relationships. He/she may already have attained a level of sexual maturity and experience in life, and may even be in a more or less stable long-term relationship (possibly with children). Therefore the question turns on what effect living with ataxia will have on the relationships that are already there. The effect of late-onset ataxia may be, ultimately, to reverse the ‘normal’ roles and polarities within a relationship, causing enormous, possibly terminal, strain to it. Ataxia Groups can offer much help, advice and support to ataxic people undergoing this particularly intense late-onset trauma.

Conclusion: understanding ataxia

To intervene effectively against the ataxias and their psychosocial consequences, it is first necessary to understand the nature of those consequences. To do this we need to construct a model of ataxia as it affects personal development at all stages; from childhood to adulthood. This is the purpose of the present article.

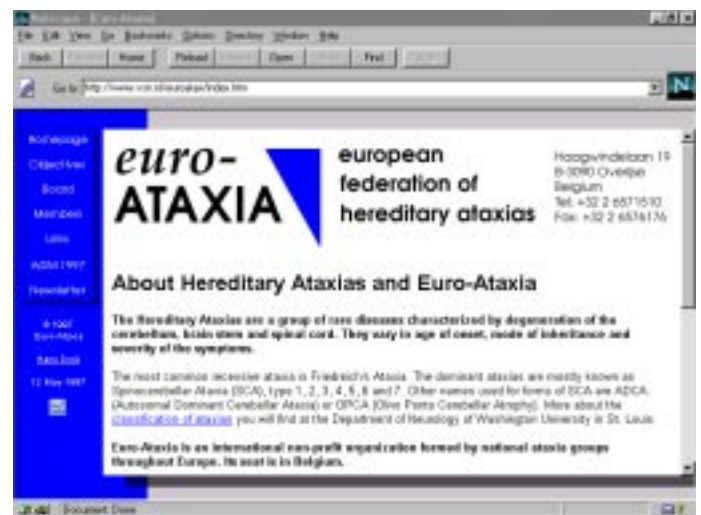
Models are not reality, nor are they intended to be so. Rather they are analytical frameworks within which questions can be addressed and interventions planned. The present chapter may leave some with the impression that all ataxic people’s experiences in life are overwhelmingly negative – ‘better never to have been born’. This, however, is to misconstrue model for reality. Most ataxic people do indeed lead rich and meaningful lives, within the constraints imposed by ataxia of course. One of the main difficulties that arose in the construction of the chapter was how to build a single psychosocial ‘model’ for ataxia. I attempted to do this by organising material around specific ‘stages of development’ in an overall ‘ataxic career’: childhood, adolescence, adulthood, etc. But this concept of ‘ataxic career’ is of course an abstract one. Individuals may recognise themselves (or parts of themselves) at certain points in the chapter but not in the whole. It is not, nor is it intended to be, a purely descriptive exercise. Its purpose is, rather, to help ataxic people themselves and those most closely involved with them – medical and welfare professionals, family and friends – make sense of and understand what is going on at this crucially important level of life. For only through such understanding can effective intervention be made: ‘He who wants to change the world must first see it as it truly is’.

ATAXIA.NET

EURO-ATAXIA WWW-pages

Recently the EURO-ATAXIA site on the World Wide Web (www.vsn.nl/euroatax/index.htm) has been completely restyled. The pages offer apart from general information on EURO-ATAXIA and hereditary ataxias, links to medical and neurological information, to the Websites of other ataxia groups, to personal Webpages of ataxic persons (send those URL’s to me!), and an online version of this Newsletter.

We will also provide you with the latest information on the coming AGM.



Ataxia groups on the Web

More and more EURO-ATAXIA members are building their own Website. Since the November 1996 issue of *Euro-Ataxia* the Finnish, German and Italian groups and the Dutch ADCA-Association opened their sites.

- **www.ms-liitto.fi**
Finnish MS Society: main page in Finnish, but also information in Swedish and English
- **ourworld.compuserve.com/homepages/DHAG**
DHAG: only in German
- **www4.iol.it/associazioni/aisa**
AISA: only in Italian
- **home.pi.net/~ataxia**
AVN: information in Dutch and English

INTERNET RELAY CHAT

Jon Bunnig

Help, I am addicted! Not to drugs or alcoholics, but since a few months I spent too much time on the so called *chatting* on the Internet. I don't damage anyone by this new hobby, but even other people do now notice my red eyes caused by less sleep. Sometimes I am so enthusiastic that I promise things to do I forget I did, like writing this article...

E-mail and discussion-groups are very useful to communicate on your PC, but sometimes, as Internet-users we want to make contact *directly*. Of course you can do this over the phone, but more often you will have extremely fun if you can talk to someone else you can not see or even you do not know! I think chatting on the Internet is totally different than the Internet itself: so if you do not like the Internet or you think it is too difficult for you, even than you can have a lot of fun by chatting.

A good method for direct communication between Internet-users is by *Internet Relay chat (IRC)*. IRC offers the possibility to talk in groups (2 or more persons). To make a comparison with the telephone: IRC offers a kind of chatter-box.

IRC doesn't have only one, but it consists of several chatter-boxes. The so called *IRC-channels*. In each IRC-channel people are talking about a certain subject, like politics. Normally I will choose for the IRC-channel #ATAXIA. This channel has initiated by some people of the National Ataxia Foundation (NAF, America). Like the name already suggests: the people who are talking on this channel has been affected by a kind of ataxia, mostly FA. Of course we do not speak only about wheelchairs and illness on this channel, but for some reason you have something in common with people over the whole world and that makes you one. After connecting the IRC-server you can choose for example for #ATAXIA and from that moment you

can follow the conversation of the other members of #ATAXIA and they will enjoy your typing.

A lot of IRC-servers exists, but it makes no difference which server you will contact: each IRC-server has connected by each other and they will exchange their messages.

So if a user selects a certain IRC-channel he will get all messages of the channel, also the messages of the users who has connected by an other IRC-server but the same channel. Examples of IRC-servers: EU.UNDERNET.ORG (Europe) or US.UNDERNET.ORG (USA).

The number of IRC-channels is varying: everyone can create a new IRC-channel. So if you are the first one selecting channel #ATAXIA, automatically you will create this channel and you will become the *channel-operator*. The channel-operator has some special rights, like removing 'undesirable' members. Do not be afraid for that it will be clear in practice. As already told #ATAXIA has been initiated by our American friends and as yet most chatters are from America, Canada, Australia and Japan. Because of the time difference you will notice that this channel will be visited frequently only after around 23.00 GMT or even later, but no-one will keep us from starting a bit earlier in Europe ! I do not know when this article has published but I also wrote this article for the Dutch newsletter and I expect a lot of new Dutch and other European chatters at around 21.00 GMT.

Although I am not an *expert*, but only someone who enjoys the chatting on Internet a lot, I am willing to answer any questions if possible. You can send me your questions by sending an e-mailmessage to: <bunnig@pi.net> or even better by writing a note in 'Myo-café' on the Dutch VSN web-site (**www.vsn.nl**) with as subject: #ATAXIA.

See you at #ATAXIA

**CLOSING DATE FOR
THE NEXT ISSUE**

1 NOVEMBER 1997

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